

What is claimed is:

Claims

1. A substantially pure human nucleic acid comprising at least 40 nucleotides that
5 hybridizes under high stringency conditions to a sequence found within the nucleic acid
of SEQ ID NO:1.
2. The nucleic acid of claim 1, wherein said sequence has a sequence
complementary to at least 50% of at least 60 contiguous nucleotides of the nucleic acid
10 encoding the methionine synthase polypeptide, said sequence sufficient to allow nucleic
acid hybridization under high stringency conditions.
3. The nucleic acid of claim 1, wherein said nucleic acid comprises a mutation or
a polymorphism, wherein said nucleic acid probe detects a mutation or polymorphism
15 selected from the group consisting of D919G, H920D, and Δ Ile881.
4. The nucleic acid of claim 3, wherein said sequence of said nucleic acid
comprises the cobalamin binding domain of the human methionine synthase gene.
- 20 5. The nucleic acid of claim 2, wherein at least 18 contiguous nucleotides of said
sequence are complementary to at least 90% of the corresponding nucleotides of the
nucleic acid encoding the methionine synthase polypeptide.
6. The nucleic acid of claim 1, wherein said high stringency conditions comprise
25 hybridization in 2X SSC at 40°C.
7. A substantially pure human nucleic acid, wherein the sequence of said nucleic
acid is at least 75% identical to the corresponding region of at least 50 contiguous base
pairs of the nucleic acid of SEQ ID NO:1.

8. A substantially pure human nucleic acid, wherein the sequence of said nucleic acid is at least 35% identical to the corresponding region of at least 50 contiguous base pairs of the nucleic acid of SEQ ID NO:1.

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9. A kit for the analysis of a human methionine synthase nucleic acid, said kit comprising a nucleic acid probe useful for detecting in the nucleic acids of a human a mutation or polymorphism in said methionine synthase nucleic acid, wherein said mutation or polymorphism is selected from the group consisting of D919G, H920D, and Δ Ile881.

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10. The kit of claim 9, wherein said probe comprises at least 40 nucleotides that hybridizes at high stringency to a sequence found within the nucleic acid of SEQ ID NO:1.

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